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Aicardi Syndrome: A Rare Case Report

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Abstract

Introduction: One case of Aicardi syndrome occurs out of every 110.000 live births, making it one of the rarest genetic neurodevelopmental disorders. Infantile spasms, agenesis of the corpus callosum, and pathognomonic chorioretinal lacunae make up the clinical triad of Aicardi syndrome. Nearly all of the patients have serious physical and cognitive impairments.

Case Report: We report a case of 8-month-old hypotonic child with low set ears, microcephaly, dolichocephaly and micropthalmia ,with bilateral iris colobomas. She was severely stunted and severely underweight with gross developmental delay. On fundoscopy, in both eyes there were characteristic well defined, circular, hypopigmented chorioretinal lacunae varying in size not involving the macula. Upon following findings, a Magnetic resonance imaging of the brain was performed which revealed agenesis of the corpus callosum and septum pellucidum.

Discussion: Aicardi syndrome represents a diagnostic challenge due to its rareness, and a multidisciplinary approach is needed to confirm the disease. Imaging plays an Important role in diagnosing aicardi syndrome. This condition is also accompanied by a variety of costovertebral, ocular, and cognitive problems.Thus, the diagnosis of Aicardi syndrome necessitates a multidisciplinary approach with opthalmology, radiology, and pediatric neurology.

Conclusion: Aicardi syndrome is a rather uncommon neurological condition. However, as seen from our study and other case report, it does exist in our nation. The median age of survival for this illness is estimated to be 18.5 years, with respiratory difficulties brought on by hypersecretion being the most common cause of mortality.

Keywords: Aicardi syndrome, Neurodevelopmental disorder, Agenesis of corpus callosum, Chorioretinal lacunae, Infantile spasms, Magnetic resonance imaging, Electroencephalogram

Introduction

Aicardi syndrome occurs in one in every 110,000 live births, making it one of the rarest genetic neurodevelopmental disorders [1, 2]. Infantile spasms, agenesis of the corpus callosum, and pathognomonic chorioretinal lacunae make up the clinical triad of Aicardi syndrome. Nearly all of the patients have serious physical and cognitive impairments.It is a rare genetic illness that was initially recognised by Jean Aicardi in 1965. Although the gene responsible for the disorder has not been found, multiple data suggest that an X chromosome gene may be responsible [3]

The incidence of recurrence in siblings is estimated t o be less than 1%, and parent-to child transmission has not been documented.We present a case of Aicardi syndrome in an 8-month-old girl. The corpus callosum was completely absent on

magnetic resonance imaging, and chorioretinal lacunae were discovered in both eyes during ophthalmoscopy. Her developmental milestones were behind schedule for her age. Aicardi syndrome usually manifests in the first 3 months of life as infantile spasms.

Case Report:

An 8-month-old female, a known case of severe acute malnutrition with global developmental delay, was brought to the casualty with complaints of fever, cough and cold. In view of having respiratory distress, she was immediately admitted to the paediatric intensive care unit for further management. Upon eliciting detailed history, relatives gave a history of hospitalisation at 5 months of age in view of failure to thrive ,seizures and global developmental delay and also NICU admission for 23 days after birth in view of being preterm (32 weeks) with low birth weight (1900 g). No history of jaundice, fever or convulsions was present during this period.She is a second born child to a non-consanguineous marriage, with maternal age being 30 years at conception.

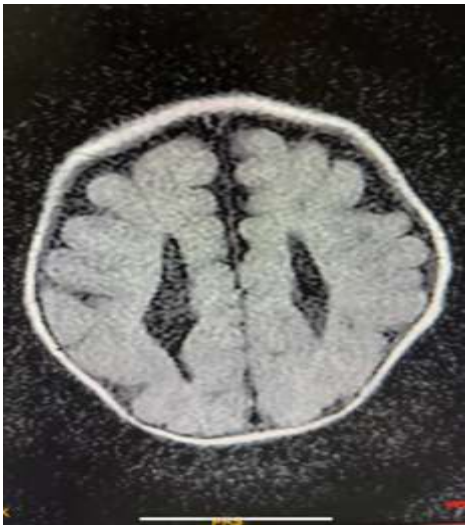
Clinical examination revealed a hypotonic child with low set ears, microcephaly, dolichocephaly and micropthalmia ,with bilateral iris colobomas. (**Fig.1.**) She was severely stunted and severely underweight with gross developmental delay. On ocular examination, the child showed micropthalmia , bilateral inferior iris colobomas and poor visual fixation. On fundoscopy, in both eyes there were characteristic well defined, circular, hypopigmented chorioretinal lacunae varying in size not involving the macula. Otoacoustic emission study was also done which was found to be normal for both ears.

Upon coming across the following findings, a Magnetic resonance imaging of the brain was performed which revealed agenesis of the corpus callosum and septum pellucidum. Bilateral colpocephaly, shallow sulci and poly microgyria was also noted. (**Fig.2**.) An Electroencephalogram was also done which showed abnormal sleep medium to high amplitude arrhythmic theta-delta waves.There was no family history of similar disorder.

Patient was referred to a paediatric neurologist and appropriate management with supplements and anti-epileptics was started.She was advised regular physiotherapy and diet counselling was done to meet her adequate nutrition requirements.Regular follow ups were also advised to keep track of her growth and disease progression.

Fig.1.

Fig.2.



Discussion:

Aicardi syndrome is difficult to recognize due to its rarity, and it requires a multidisciplinary approach to diagnose the disease.

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Imaging plays an Important role in diagnosing aicardi syndrome.Prenatal ultrasound results may be suggesti ve of Aicardi syndrome, but they cannot be used to di agnose it. Aicardi et al. stated that the existence of choroid plexus cysts and agenesis of the corpus callosum are strongly suggestive of Aicardi syndrome[4]. It can be associated with partial or complete corpus callosum agenesis.In this patient's instance, complete corpus callosum agen esis was observed [5].Other ocular abnormalities reported are optic nerve colobomas, optic nerve hypoplasia, optic disc pigmentation, microphthalmos, retinal detachment, macular scars, cataract, iris synechiae and iris coloboma which was seen in our case.[6]Costovertebral malformations such as fusion of vertebrae, kyphoscoliosis, absent ribs, and occasionally cleft lip and palate may also be associated with Aicardi syndrome.[7]

The developmental delay in Aicardi syndrome is generally profound, involving both motor and language skills.This condition is also accompanied by a variety of costovertebral, ocular, and cognitive problems.Thus, a multidisciplinary approach involving paediatric neurology, radiology, and ophthalmology is required for the diagnosis of Aicardi syndrome. Treatment is individualised and long-term management is needed, along with physical therapy, occupational therapy, and speech therapy. Surveillance includes routine monitoring of growth, nutritional status, seizure control and developmental progress.

Conclusion:

Aicardi syndrome is a rather uncommon neurological condition. However, as seen from our study and other case reports [9,10,11,12], it does exist in our nation. The median age of survival for this illness is estimated to be 18.5 years, with respiratory difficulties brought on by hyper-secretion being the most common cause of mortality [13,14].

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